

REVIEW ARTICLE

The geneticists clinicians consortium of India: an initiative to bridge the gap between genetics and clinical medicine

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ABSTRACT

The Geneticists Clinicians Consortium of India (GCCI) is a consortium whose aims are to establish a knowledge of clinical and genomic data, thereby bridging the gap between clinicians and geneticists. The broad principle of the GCCI is to connect the geneticists and clinicians of India and promote high standards in genetic counseling with the help of our qualified health care team and certified genetic counselors. In keeping with this, the principal objectives of the GCCI are to support the dissemination of information from scientific research and create awareness to enhance the profession of genetic counseling in India.

Keywords: Genetics, clinicians, mutations, health, awareness.

Introduction

The Geneticists Clinicians Consortium of India (GCCI) aims to promote high standards in genetic counseling by bridging the gap between geneticists and clinicians in India, supported by our qualified healthcare team and certified genetic counselors. Our core objectives are to disseminate information from scientific research, create public awareness, and enhance the profession of genetic counseling across India. GCCI also facilitates mentor-mentee relationships through educational programs that focus on the basics of genetics, prenatal testing, and newborn screening, promoting interaction and expertise sharing. We are organizing an outreach program for clinicians, young clinical researchers, scientists, and life science graduate students interested in genetics. The goal is to empower young researchers with both clinical and genetic information concerning causative genes, disease diagnosis, and available treatment options for single-gene diseases, complex genetic disorders, and birth defects. Additionally, newborn screening for sickle cell disorders has been initiated in several regions of India through efforts led by the Indian Council of Medical Research and the National Rural Health Mission of various states to enable advancement. Over the years, an imperative need has been felt to check prenatal diagnosis and genetic counseling in Asia (1-3). We describe the development and aims of the GCCI initiatives.

The main guiding goals of GCCI are to promote the science and practice of Human Genetics in India:

- To promote scientific collaborations in human genetics, genomics, and allied scientific disciplines
- To impart education on specialized topics in human genetics, genomics, and allied scientific disciplines through the organization of summer and winter schools, workshops, symposia, conferences, meetings, and so on.
- To promote public understandings on human genetics, genomics, and allied scientific disciplines
- To publish periodic newsletters, articles, and so on, for enhancing communication among scientists and community outreach
- To serve as a vehicle for interaction of human geneticists with patient collation groups, funding agencies, clinical and scientific societies of relevance, and so on, for more effective conduct of scientific

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Received: 10 September 2025 | **Accepted:** 24 January 2026



research and translation in human genetics, genomics, and allied scientific disciplines

- To undertake projects for the enhancement of basic understanding of genomics and health, and for genomics-based improvement of human health and welfare
- To bridge the gap between geneticists and clinicians in bringing an effective collaborative groups.

The consortium includes a committee to check genomic/clinical data and provide quality control for counselors/executive committee to introduce and review the proposals for conducting courses and workshops to address key issues pertaining to genetic disorders, complex disorders relating to genotype-phenotype correlation. The coordination between clinicians and geneticists requires better education and training for both groups. The clinicians need to be provided with more genetics education and training to increase their comfort and confidence in interpreting genetic test results and incorporating them into clinical decision-making. The geneticists need to better understand the clinical implications of genetic findings and be able to communicate these findings in a way that is easily understood by clinicians and patients. In addition, there needs to be increased collaboration between clinicians and geneticists, with genetic counselors and other specialists playing a key role in the integration of genetics into clinical practice, which has the potential to improve patient outcomes and advance the field of precision medicine. We set up a vision 2030 to prioritize standard operating procedures to close the gap between clinicians and geneticists, as we unlock the full potential of genetics to transform the way we diagnose and treat disease.

Organization and Background

The GCCI initiative was taken jointly by Bioclues (<https://bioclues.org/> last accessed on December 1, 2025) and Aabir Bio-Services Foundation (ABSF: <https://www.bio-services.org/> last accessed on December 1, 2025) in 2020. At GCCI, we want to foster healthy connections between clinicians and geneticists. In 2005, Bioclues was founded with a view to serving as a hub for mentor-mentee relationships. Over the years, this expanded further and several initiatives were taken to create and disseminate information both basic and recent, through our online platform and webinars. In due course, GCCI was established in collaboration with ABSF with a mission to facilitate knowledge transfer and raise awareness with regard to preventive healthcare with a focus on women and girls. Besides the prominent inherited genetic conditions, Aabir Bioservices also recognizes the increasing burden of environment and lifestyle-induced acquired genetic disorders, deteriorating reproductive health amongst the population, and sexually transmitted diseases in women and are committed to addressing the associated health issues.

Through GCCI, our main focus has been to establish an alliance between clinicians and geneticists to make each aware of the contribution of the other and ensure the quality service to real people, especially in

underprivileged communities. Now with the current program, we aim to inspire, motivate, and guide students, Figure 1 post-grads, practicing physicians, and genetic counselors. By the end, we intend to empower both ends of the rope; clinicians and geneticists to be able to prescribe the best medical option advice, both diagnostic and therapeutic, as well as to keep in sync with the patients. Over the last 5 years, we organized four workshops and as many as 38 webinars with the most recent being held at ICMR-NIRTH (<https://www.nirth.res.in/event.phplast> last accessed on November 25, 2025). While the events were largely in lieu of Applied Genetics and Genetic Counseling, we have over 80% satisfactory feedback from participants.

Clinicians' perspective and genetic counseling

The GCCI intends to raise public awareness of prevalent genetic diseases present in that area, such as sickle cell anemia, thalassemia, and Down syndrome. We aim to educate the general public on how to recognize genetic problems in their families, as well as medical professionals on when and where to refer suspected genetic patients. By promoting scientific activities to the general public, we hope to strengthen the educational programme and close the gap between doctors and clinicians. Sheldon Reed first used the term "genetic counseling" in 1948 as it aids individuals in comprehending and adjusting to the medical, psychological, and familial ramifications of genetic anomalies and illnesses (4). Being a subjective art, genetic counseling has experienced its fair share of differing opinions, disputes, and squabbles throughout the years. Genetic clinics were first established in wealthy nations in the 1940s. However, a lot of these clinics supported the eugenics agenda, which aimed to manage population growth overall while limiting the rise of particular races and ethnic groups. This was swiftly replaced with a non-directive strategy, enabling patient autonomy. Prior to recently, the majority of patients heavily relied on their primary health care provider for information about problems related to their condition. However, the majority of medical professionals were not always aware of multiplicities of the variety of syndromes and their genetic relationships, as well as the diagnostic and prognostic genetic tests that were accessible. With the advent of genetic counselors specialists who are extremely specialized in genetics and counseling, the focus of modern genetic counseling was shifted in the twenty-first century. These genetic counselors now provide people with more functional information, which is non-directive therefore patients get the chance to make an informed decision. Additionally, it has enabled patients, lab workers, and medical professionals to join hands together. The profession continued to grow as a result of improvements in diagnostic methods and genetic information advancements. Genetic counseling is now more tailored than just communicating, taking into account a patient's history, their family tree, how inheritance is explained, and the likelihood of recurrence (5). The American Society of Human Genetics and European Society for Human Genetics (ESHG) have rigorous education programs on genetics but not on genetic counseling. As educational resources for the



Figure 1. Organizational framework of GCCI.

Human Genetics Community they contain information on courses organized by the ESHG, with a webcast archive, while we at bioclues.org/gcci have YouTube pages at youtube.com/biocluesin where all recorded events are inserted from time to time.

Genetic counseling is now more accessible, routed, and multifaceted. Couples planning a pregnancy, especially those from ethnic groups where recessive genetic disorders are common, can benefit from the advice of genetic counselors. In wealthy nations, screening tests for pregnant women, especially those over 35, have practically become commonplace, enabling counselors to estimate risks. Similarly, prenatal genetic counseling can address worries about characteristics that might influence the baby, particularly in couples who have a history of infertility, stillbirth, or numerous miscarriages. Within the first 72 hours following birth, even newborns are checked and counseled for genetic disorders (newborn screening), ensuring prompt and effective disease management. But the situation in India is still very different and challenging (6). Since there is no government assistance available and few health insurance plans cover these fees, genetic testing and counseling must be prescribed with caution. As a result, testing and counseling are directly impacted by cost limits. In addition, with the development of newer technologies, such as clinical exome and whole genome sequencing, a vast amount of data is produced, and there are difficulties in interpreting polymorphisms and variants

of uncertain significance (VUS). This has also resulted in ethical, psychological, and socio-medical issues, such as the question of whether or not to communicate with patients on late-onset disease or a predisposition to other diseases that is obvious on the test results. We predict that the practice of counseling will transform and become more diverse as new genetic, biochemical, and molecular testing are developed. It will enable precise interpretation and trustworthy treatment and preventative methods. We also expect that patients would speak with counselors directly in cases of congenital defects, unusual test results that point to a genetic illness, chromosomal disorders, poor obstetric history, repeated miscarriages, stillbirths, multiple abortions, and similar situations alike.

Phenotype-genotype correlation

Phenotype characterization and validation have been essential components of genetic analysis since Gregor Mendel employed the visible features of pea plants to identify and track units of inheritance. There are several reasons why phenotypes are significant. First, they provide genetic modification and allow us to see features and occurrences that are genetically inherited. Second, it has successfully taken advantage of genetic mutations that give a growth or survival advantage or a characteristic that could be physically measured. Third, although they represent the manifestation of genotypes and show gene function, phenotypes are very important.

In this sense, phenotypes represent a crucial step between fundamental genetics and biological comprehension (7). Understanding the mechanism of such a genotype-phenotype link became feasible because of the molecular transformation in biology. The recent advent of deep mutational scanning assays, which can score extensive genotypic libraries for health and several phenotypes in an enormously parallel fashion, is made possible by breakthroughs in DNA synthesis and sequencing (8).

Genome-wide Association Studies (GWAS) have been of great help in recognizing various risk-associated loci in several diseases, as their causal molecular mechanisms are studied from time to time. For such genetic characterization, genetically modified mice are being used (9). Such studies work under the theory that the genotype-phenotype relationship of a mouse can be generalized to humans. The mouse is the first mammalian model organism. The arrival of nucleases aided genome editing strategies such as CRISPR/Cas9 has raised the use of mice for genotype-phenotype relationship studies. Without any contradiction, we can say that mice's genetics are making valuable contributions to reproducibility and helping medical sciences. Consortium like International Knockout Mouse Consortium is established to create knockout mice lines to study the phenotypes (10).

Several genetic mutations are involved in genetic interactions that would interrupt phenotypes in different ways. By studying model organisms, we translate it for other systems to establish genotypic-phenotypic relationships. Besides this, predicting any disease heritability remains a major challenge in modern genetics. Next generation sequencing on the other hand along with the electronic health records and population-centered registries, has helped maintain the directory of millions of human variants, which are also an excellent source of information for understanding genotypic and phenotypic relationships concerning human abnormalities (9). The GWAS has provided the advantage of this huge data in comparing the variants with different phenotypes (11). Even after this linking, there remains a disparity in disease risk prediction and chances of heritability based on familial aggregation. This disparity could be explained based on "missing heritability" or different combinations of either common or rare variants that play a role in phenotype inheritance and susceptibilities out of genetic interactions of variants (12). Due to the enormous number of potential gene-gene combinations inside the human genome, most approaches sadly lack the statistical capacity to discover genetic interactions in large-scale genotyping datasets (13). For instance, breast cancer cohorts with genetic interactions between combinations of human pathways are linked to an increased or decreased risk of illness (14). In the end, comprehensive mapping of genetic interactions in model human cell lines will provide a worldwide genetic network for various human cells, giving a powerful data-driven resource that defines a functional wiring schematic for humans, as done with yeast. The systematic discovery of pathway-level genetic interactions from human genotyping data should be made possible by utilizing genetic interactions to define functional modules and map their relationships. This will result in a deeper understanding of human biology and

improve our understanding of the relationship between genotype and phenotype (15). The quantitative genotype-health fitness maps open the door to the predictive models, possibly enhancing our capacity to manage the mode of action of any pathogen or malignant cells with the help of sequencing data (8).

Understanding individuals' genetic differences and their role in preventing or predisposing people to disease is crucial for providing individualized healthcare. With data obtained from the clinical trials and the biomedical literature, several efforts seek to provide this information by curating manually such genotype-phenotype connections in structured databases. However, delays in updating such databases with the most recent information have been caused by the exponential growth of the biomedical literature and the limited capacity of manual curators to locate the genotype-phenotype associations "hidden" in text. As a result, it is difficult to use the useful information that is now accessible to create customized healthcare solutions (16).

GWS as a guide for genetic counseling

The most accurate technique currently available for identifying harmful genetic variations is GWS. Best clinical practice dictates that GWS be administered as a comprehensive clinical service that includes both pre-test and post-test genetic counseling following North American and Europe regulations. In 50% of patients with significant but nonspecific diseases, GWS of exomes or entire genomes can pinpoint a particular hereditary or de novo genetic etiology. While cytogenetic analysis and chromosomal microarray analysis, the prior clinical standards for genome-wide testing for genetic origins of illness, are still useful, GWS offers a significant boost in diagnostic sensitivity. GWS's main strength is its capacity to detect single-nucleotide variations and small- or large-copy number variations anywhere in the genome, regardless of the genes or regions implicated. Our genomes diverge from the human reference genome by 4–5 million nucleotides. Therefore, determining one or two pathogenic variants from a vast pool of largely benign variations that are unlikely to be connected to the patient's ailment is required to interpret GWS data. GWS differs from the majority of other clinical tests, including targeted genetic testing, due to the intrinsic difficulty of the challenge, which frequently pushes us to (or beyond) the limits of our present understanding of genome biology. GWS often reveals unclassified variants or VUS, secondary findings (which can indicate multiple and unexpected disorders), and difficulties associated with diagnosing rare diseases, in contrast to a conventional single gene or chromosomal genetic counseling.

Additionally, in up to 5% of patients receiving GWAS, additional genetic diagnoses that affect their clinical phenotype will be found. Further, when the patient's condition changes from time to time or as the phenotypic spectrum linked to such variations is elucidated in the medical journals, certain VUS may later be designated as disease-causing. Similar to how variations originally identified as disease-causing might eventually be reassigned as benign or having unknown relevance.

These reclassifications may significantly impact families and healthcare professionals. By proving that the patient's situation is being addressed with continued care, variant reclassification can increase confidence between a genetic counselor and the patient. However, the uncertainty brought on by a VUS might cause psychological difficulties. For this reason, genetic counseling for families thinking about GWS prepares them for the unforeseen. Genetic counseling helps prepare families when a diagnosis is a gray since most patients and doctors view diagnoses as either true or false. Furthermore, genetic counseling is crucial when employing a test that might push the boundaries of our clinical and scientific understanding (17,18).

Scientists' perspective

Genetic counseling is now delivered by a variety of medical and allied health experts (clinical and human geneticists, physicians with continuing education in genetics, and medical social workers), necessitating the development of a standardized method of genetic counseling. Graduates trained in India are increasingly entering the genetic diagnostics sector in jobs such as genetic counseling, test reporting, sales, marketing, and medical science liaison. Genetic counselling education in India has grown considerably in recent years, with structured training pathways now available across multiple academic levels. Formal postgraduate degree programs, including the M.Sc. in Genetic Counselling at Kasturba Medical College (MAHE, Manipal), the M.Sc. in Genomic Counselling at MAHE Bengaluru, and the M.Sc. in Genetics and Genomic Counselling at DY Patil University (Mumbai), form the core of academic preparation for future genetic counsellors. In parallel, several postgraduate diploma programs - such as those offered by the Institute of Genetics and Hospital for Genetic Diseases (Osmania University, Hyderabad), JIPMER Puducherry, and MDU Rohtak - provide specialized training for graduates and clinicians seeking to expand competencies in genetic counselling. Additionally, short-term certificate programs and clinical genetics workshops are delivered by institutions including St. John's Medical College (Bangalore) and SGP GIMS (Lucknow), as well as by private and university-based providers, supporting continual professional development for healthcare professionals in the following areas:

- a. Cancer
- b. Cardiology
- c. Diabetes
- d. Neurology/Mental Health
- e. Prenatal Care, Newborn Screening, and Reproductive Health

Conclusion and Future Outlook

The GCCI represents a collaborative effort to integrate genetics into routine clinical practice in India. Continued expansion of training programs, standardized counseling protocols, and policy-level engagement will be essential to sustain its impact. Genetic counseling is a burgeoning profession around the world, and international collaboration and reciprocal agreements are helping

to advance training, regulation, and practise scopes. Versatility is a key quality that would benefit Indian genetic counselors. Through GCCI, we firmly believe and hope that genetic services could be taken up to the next level with proactive steps, thereby setting professional guidelines for clinical geneticists and genetic counselors. This would be a significant step toward the country's establishment of a community genetic service paradigm. To properly manage expanding technology, novel genetic testing, diagnosis, advocacy, and appropriate referral, clinical geneticists and genetic counselors must broaden their area of practice. There is an impending need to join hands. Together we can!

List of Abbreviations

GCCI	Genecists Clinicians Consortium of India
VUS	Variants of Uncertain Significance
ABSF	Aabir BioServices Foundation
ASHG	American Society of Human Genetics
ESHG	European Society of Human Genetics

Clinical trial number

Not applicable.

Conflict of interest

The authors declare that they have no conflict of interest regarding the publication of this manuscript.

* The GCCI members at <https://bioclues.org/gcci> (Kausar Neyaz, Shalini Rajagopal, Dhanlakshmi Shetty, Sugunakar Vure, Obul Reddy Bandapalli).

Funding

None.

Consent to participate

Not applicable.

Consent for publication

The authors provide consent to publish the manuscript.

Ethical approval

Not applicable.

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